

**OXFORD CAMBRIDGE AND RSA EXAMINATIONS**

**Advanced GCE**

**HUMAN BIOLOGY**

Genetics, Homeostasis and Ageing

**2867**

Tuesday

**31 JANUARY 2006**

Afternoon

2 hours

Candidates answer on the question paper.

Additional materials:

Electronic calculator

Ruler (cm/mm)

Candidate Name	Centre Number	Candidate Number												
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**TIME** 2 hours

**INSTRUCTIONS TO CANDIDATES**

- Write your name in the space above.
- Write your Centre number and Candidate number in the boxes above.
- Answer **all** the questions.
- Write your answers, in blue or black ink, in the spaces provided on the question paper.
- Read each question carefully before starting your answer.

**INFORMATION FOR CANDIDATES**

- The number of marks is given in brackets [ ] at the end of each question or part question.
- You will be awarded marks for the quality of written communication where this is indicated in the question.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.

FOR EXAMINER'S USE		
Qu.	Max.	Mark
1	14	
2	17	
3	18	
4	22	
5	15	
6	20	
7	14	
<b>TOTAL</b>	<b>120</b>	

**This question paper consists of 20 printed pages.**

Answer all the questions.

1 Osteoarthritis and osteoporosis are both conditions which may affect the skeletal system as it ages.

(a) Fig. 1.1 shows the changes which occur in the knee joint, in an individual with osteoarthritis .

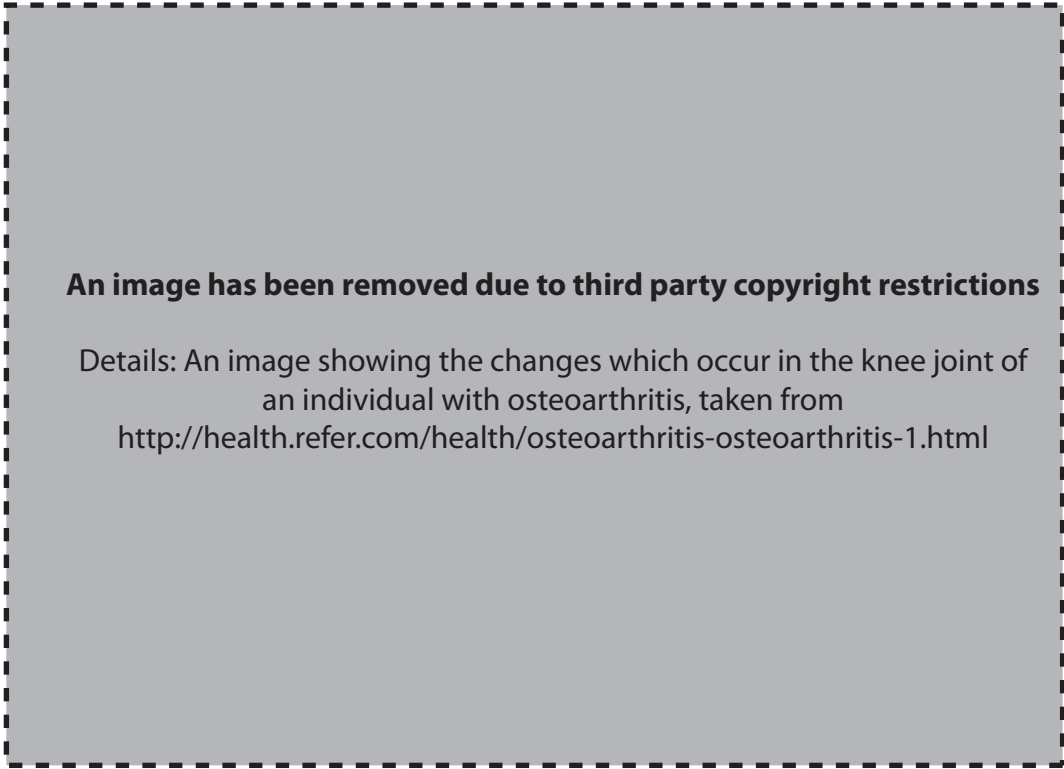


Fig. 1.1

(i) Describe the changes in A, B and C that are associated with osteoarthritis.

A .....

B .....

C .....[3]

(ii) Explain why these changes cause pain.

.....  
.....  
.....  
.....[2]

(iii) Osteoporosis is another serious bone disease.

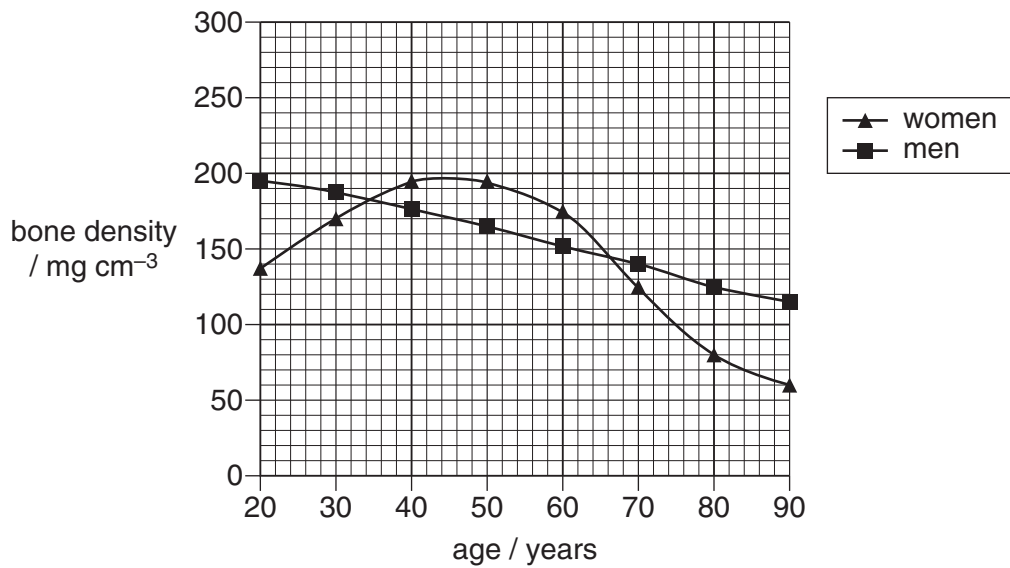
Describe the main **differences** found in structures **A** to **C** on Fig. 1.1, if the knee joint was damaged by **osteoporosis**.

**A** .....

**B** .....

**C** .....[3]

(b) Fig. 1.2 shows the changes in bone density in men and women as they age.



**Fig. 1.2**

(i) Using the data in Fig. 1.2, describe how bone density changes with age.

.....  
 .....  
 .....  
 .....  
 .....  
 .....  
 .....  
 .....  
 .....  
 .....[3]







(b) One cause of ageing involves changes in the chromosomes.

The ends of the chromosomes are protected by identical repeating lengths of DNA called **telomeres**.

These prevent the chromosomes unravelling during cell division, in a similar way to the plastic end on a shoelace that prevents the shoelace unravelling.

Each time a cell divides, the length of the telomeres shortens and the ability of the cell to divide decreases. This is part of the ageing process.

(i) Suggest why it may be an advantage if 'the ability of the cell to divide decreases' .

.....  
.....[ 1]

(ii) Some cells contain high levels of an enzyme called telomerase. This enzyme prevents the telomeres shortening by adding more repeating units of DNA to the telomere. Cells with high levels of telomerase therefore continue to divide.

Fig. 3.1 shows the length of the telomeres in cells dividing to form two different kinds of tissue.

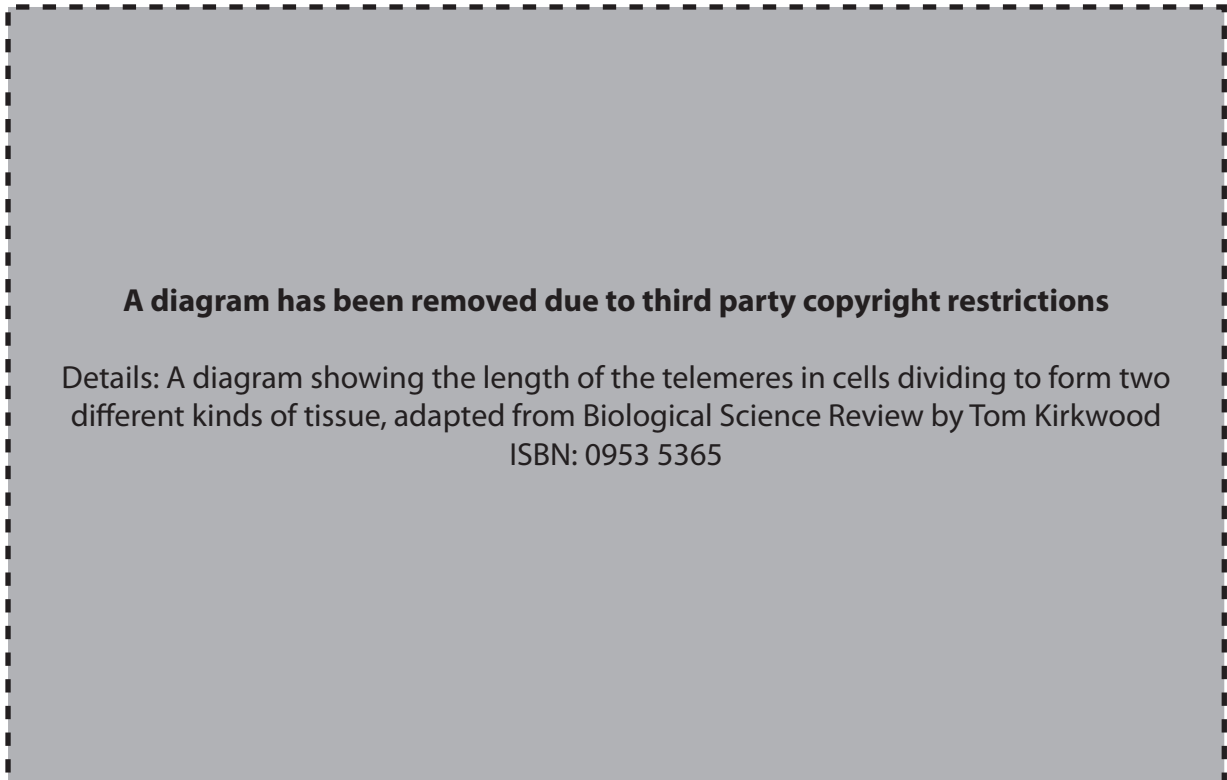


Fig. 3.1

Suggest an example of tissue A and tissue B.

tissue A .....

tissue B .....

[2]









5 Haemophilia A is a genetic disease in which the blood does not clot. It is caused by the absence of a protein called Factor VIII.

(a) Show, **by means of a genetic diagram**, the probability of a son inheriting haemophilia from a mother who is a carrier and a father who is not haemophiliac.

*key*

*parental genotypes* .....

*gametes* .....

*probability* .....[5]

- (b) Individuals with haemophilia may have uncontrolled bleeding from a minor wound.

Suggest how an ice pack would help to control the bleeding from the wound.

.....

.....

.....

.....

.....

.....

.....

.....[3]

- (c) If haemophiliacs play contact sports they may bruise any part of their bodies. The resulting damage may cause uncontrolled internal bleeding. This is treated with injections of genetically engineered Factor VIII.

Table 5.1 shows the percentage of a full dose of Factor VIII which is needed, depending on which part of the body is bleeding.

**Table 5.1**

part of the body	dose of Factor VIII / % of full dose
elbow	35
ankle	25
muscle	35
head	100

- (i) Suggest why there is such a variation in dosage to treat internal bleeding in different parts of the body.

.....

.....

.....

.....

(ii) What might be the consequences if too little Factor VIII was given for internal bleeding from a head injury?

.....  
.....  
.....

[5]

(iii) Before genetically engineered Factor VIII was available, its only source was from donated blood.

State **two** advantages of using the genetically engineered Factor VIII.

1 .....  
.....

2 .....  
.....

[2]

[Total: 15]

6 In a population, the frequency of a normal allele (**p**), and an abnormal allele (**q**) at a specific gene locus, can be calculated using the Hardy-Weinberg equations. These are based on the Hardy-Weinberg principle.

(a) Explain what is meant by the Hardy-Weinberg principle.

.....

.....

.....

.....

.....[3]

(b) In East Africa where malaria is common, 4% of the population have sickle cell disease and are homozygous for the sickle cell allele (Hb<sup>S</sup> Hb<sup>S</sup>).

Use the two Hardy-Weinberg equations below to calculate the percentage of the East African population who have **sickle cell trait**.

$$p + q = 1$$

$$p^2 + 2pq + q^2 = 1$$

where  $p^2$  = frequency of the normal genotype Hb<sup>A</sup> Hb<sup>A</sup>  
 $q^2$  = frequency of the sickle cell genotype Hb<sup>S</sup> Hb<sup>S</sup>  
 $2pq$  = frequency of genotype Hb<sup>A</sup> Hb<sup>S</sup>

Show your working.

Answer = .....% [6]

(c) Explain why the percentage of individuals with sickle cell **disease** in the United Kingdom is much lower than in East Africa.

.....  
.....  
.....  
.....  
.....[3]

(d) Suggest why the Hardy-Weinberg equations would be useful when explaining the differences which exist between the percentage of individuals with sickle cell **trait** in the United Kingdom and the percentage in East Africa.

.....  
.....  
.....  
.....  
.....[2]

Question 6 continues on page 16

- (e) Sickle cell anaemia is caused by a gene (point) mutation in the gene coding for the  $\beta$  globin chain of haemoglobin.

The base sequence involved in the mutation is shown below in the normal allele ( $Hb^A$ ) and the mutant allele ( $Hb^S$ ).

**Normal allele ( $Hb^A$ )** .....C C T G A G G A G.....

**Mutant allele ( $Hb^S$ )** .....C C T G T G G A G.....

- (i) Explain why the gene still functions to produce a form of haemoglobin.

.....

.....

.....

.....[2]

- (ii) Explain how this mutation may cause the symptoms of sickle cell **trait**.

.....

.....

.....

.....

.....

.....

.....

.....[4]

[Total: 20]



7 A fifty year old woman visits her doctor because she is feeling generally unwell.

- She is not sleeping well due to the need to pass urine at frequent intervals during the night.
- She is always tired regardless of how much she rests.
- She is often hungry and craves sweet food.
- Occasionally she feels dizzy.

The doctor suspects that the patient may have Type 2 diabetes.

(a) (i) Describe **two** investigations that the doctor should order to confirm the diagnosis.

1 .....

.....

.....

2 .....

.....

.....[4]

(ii) Suggest what advice the doctor might give the patient to produce an improvement in her condition.

.....

.....

.....

.....[2]

**Question 7 continues on page 18**

- (b) Type 2 diabetes is usually caused by the failure of the target cells to respond to insulin due to a defect in the insulin response mechanism of the cell.

Fig. 7.1 shows the normal sequence of events when insulin reaches a target cell.



Fig. 7.1

Using the information in Fig. 7.1,

- (i) suggest the composition of the insulin receptor and explain how this may introduce a genetic risk factor for Type 2 diabetes;

.....  
.....  
.....[2]

- (ii) describe the sequence of events that increase the number of glucose channels in the cell surface membrane.

.....  
.....  
.....  
.....  
.....[3]

- (c) Further investigations revealed that the patient had a significant reduction in blood flow to her lower legs and feet caused by fatty deposits in her arteries (atherosclerosis).

Excess blood glucose is converted into fat by the liver. This fat contributes to the development of atherosclerosis.

Explain how fat from excess blood glucose may cause atherosclerosis.

.....

.....

.....

.....

.....

.....[3]

[Total: 14]

**END OF QUESTION PAPER**

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